Annals of Clinical and Analytical Medicine
Case Report

Arterial thrombosis revealing a Factor V Leiden mutation: A Case Report

Ilham Elouardighi, Lamya El Iaziji, Soukaina Batali, Amina Barkat
Departement of National Reference Center for Neonatology and Neonatal Resuscitation, Faculty of Medicine and Pharmacy, Mohammed V Souissi University, Rabat, Morocco.

Abstract

Introduction: Vascular thrombosis in newborns is rare and has a poor prognosis. Several conditions, such as sepsis and perinatal asphyxia, lead to a procoagulant state. Blood hyperviscosity is also promoted by polycythemia, acute dehydration, congenital heart disease, perinatal asphyxia, etc.

Case Presentation: We report the observation of bilateral thrombosis of the humeral, radial, and ulnar arteries with early neonatal onset to illustrate the diagnostic, therapeutic, and evolutionary aspects of this rare form of early neonatal arterial thrombosis. The diagnosis was confirmed by Doppler ultrasound, with the discovery during the etiological assessment of an authentic Factor V Factor V Leiden mutation.

Conclusion: This observation provided an opportunity to review the literature concerning the evolving clinical and therapeutic aspects of neonatal arterial thrombosis. Although there are many therapeutic options, there is no international consensus on the management of neonatal arterial thrombosis, and practices vary between centers.

Keywords

Factor V Leiden mutation, neonatal thrombosis, arterial thrombosis, neonate, doppler ultrasonography

DOI: 10.4328/ACAM.22997 Received: 17/11/2025 Accepted: 05/01/2026 Published Online: 17/01/2026 Printed: 20/03/2026 Ann Clin Anal Med 2026;17(Suppl 2):S196-199
Corresponding Author: Ilham Elouardighi, Departement of National Reference Center for Neonatology and Neonatal Resuscitation, Faculty of Medicine and Pharmacy, Mohammed V Souissi University, Rabat, Morocco.
E-mail: elouardighiilham@gmail.com P: +212 4866 3727
Corresponding Author ORCID ID: https://orcid.org/0009-0001-1728-2310
Other Authors ORCID ID: Lamya El Iaziji, https://orcid.org/0009-0000-4679-5839 · Soukaina Batali, https://orcid.org/0000-0002-2119-3521
Amina Barkat, https://orcid.org/0000-0002-5724-2751

Introduction

Vascular thrombosis is rare in newborns but is often very serious. Its occurrence is partly determined by specific characteristics of coagulation and fibrinolysis.[1] The few epidemiological studies highlight the prevalence of spontaneous renal venous thrombosis and thrombosis complicating vascular catheterization. Clinical symptoms vary depending on the location and extent of the thrombosis and the effectiveness of the bypass system. Their diagnosis is increasingly reliable thanks to imaging techniques. Arterial thrombosis of the limbs rarely occurs in utero.[2] They are serious because they can compromise functional and vital prognosis.[3-4]

We report the observation of bilateral thrombosis of the humeral, radial, and ulnar arteries with early neonatal presentation to illustrate the diagnostic, therapeutic, and evolutionary aspects of this rare form of early neonatal arterial thrombosis.

Case Presentation

This is a full-term male newborn from a poorly monitored pregnancy, born to a 31-year-old mother, G1P1 primigravida, primipara, with no known chronic conditions and no history of consanguinity, blood type AB+. The infectious history was negative, with a history of preeclampsia with oligoamnios.

The delivery was vaginal, Apgar score 9/10/10, birth weight was 2900g.

The newborn was admitted at 10 hours of life for respiratory distress rated 3-4 / 10 according to the Silverman score after a free interval. The patient was intubated and ventilated upon admission due to respiratory acidosis on blood gas analysis.

The newborn presented with sudden ischemia of the right hand and the first two fingers of the left hand, with no evidence of iatrogenic causes. Clinical examination revealed bilateral axillary pulses, but no humeral or distal pulses (Figure 1).

An emergency Doppler ultrasound was performed to confirm the absence of blood flow in the right ulnar and radial arteries and the left radial artery.

This was supplemented by an angiography scan of the upper limbs, which showed occlusion of the right humeral artery at its distal third, extending to the radial and ulnar arteries, with occlusion of the distal third of the left radial artery (Figure 2).

A crash and thrombophilia assessment was requested, showing: Hemoglobin at 16.8 g/dl, platelet count at 197,000 u/l, prothrombin time (PT) normal at 100%, quick time (QT) at 11.4 sec, patient TCA/control TCA at 1.64, normal fibrinogen level at 2.4g/l, D-dimers at 2.4ug/l, hyperhomocysteinemia at 25.74 µmol/l, total protein S at 95%, normal protein C, and Factor II activity at 88.6%, FVII at 133.7%, FVIII at 200%, FIX at 101.3%, FX at 152.1%, FXI at 163.6%.

The genetic study of the factor II mutation was negative, and the test for the factor V Leindein gene mutation was positive.

Neurologically, the patient was very hypotonic, unresponsive, with weak primitive reflexes.

An initial brain CT scan was requested, which showed no abnormalities, supplemented by a brain MRI angiogram, which indicated cortical lesions of the corpus callosum and deep white matter of ischemic appearance.

The evolution was marked by the absence of color Doppler imaging with minimal flow preservation on pulse Doppler imaging in the lower third of the left radial artery and repermeabilization of the arterial axis of the right upper limb.

The patient was placed on thrombolytic therapy while waiting for the lesions to stabilize in order to consider amputation of the necrotic part of the right upper limb, but he died in a state of refractory septic shock despite antibiotic treatment and the necessary resuscitation measures.

Family screening was carried out, and the family investigation concluded that the mother was a carrier, heterozygous for the Factor V Leiden mutation.

Ethical Approval

This study did not require ethical approval according to the relevant guidelines.

Statistical Analysis

Not applicable. This study describes a single clinical case and no statistical analysis was performed.

Reporting Guidelines

This case is reported in accordance with the CARE guidelines.

Discussion

Arterial thrombosis is rare in newborns. Several conditions,

Figure 1. Appearance of ischemia in the right hand

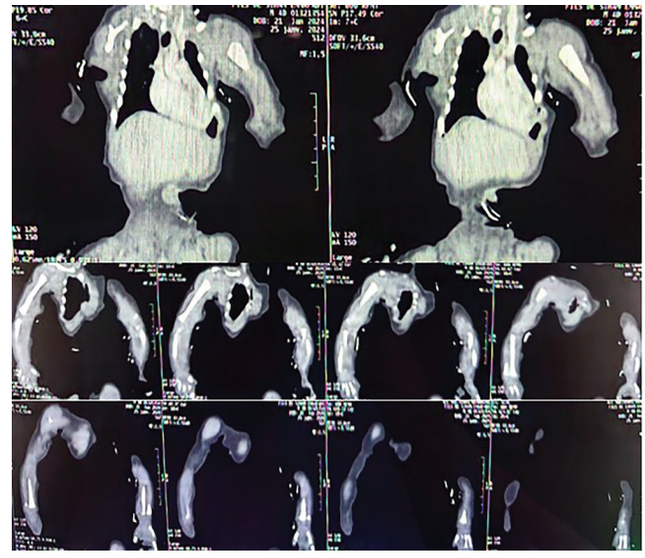

Figure 2. CT angiography images of the upper limbs showing occlusion of the right brachial artery and occlusion of the distal third of the left radial artery

such as sepsis and perinatal asphyxia, lead to a procoagulant state. Blood hyperviscosity is also promoted by polycythemia, acute dehydration, congenital heart disease, and a coagulation disorder causing thrombophilia, as in the case of our patient.[4]

Thrombotic events in newborns have been little studied. The most significant series are the Danish cohort by Tuckuviene et al. and by Monagle et al.[5,6]

The incidence of vascular thrombosis in the Danish study was 2.4%, with arterial localization in one-third of cases.[5] The sites involved were the aorta in 36% of cases and the iliofemoral arteries in half of the cases.

However, the incidence of vascular thrombosis in the prospective study by Monagle et al. was 5 per 100,000 births. Thrombosis was arterial in 19 newborns (24%), with iliofemoral localization in 8 cases (40%), cerebral in 7 cases (36%), and aortic in 2 cases (10%).[6]

The factors responsible for thrombus formation are endothelial damage, blood hyperviscosity, and coagulation disorders. Blood hyperviscosity is observed in 1 to 5% of newborns, who are mainly at-risk newborns, particularly those born to diabetic or smoking mothers and those who suffered perinatal asphyxia.[7]

In newborns, certain hereditary deficiencies in hemostatic proteins have been recognized as causing thrombosis.

In our patient, the thrombosis was secondary to resistance to activated protein C following a Factor V Leiden mutation.

Of all hereditary thrombophilias, the one linked to factor V Leiden is the most common. When this mutation is present, the level of factor V synthesis and its procoagulant activity remain unchanged, but the inactivation of factor V by activated protein C is reduced, leading to resistance to activated protein C.[8] This mutation is generally sought after because resistance to activated protein C has been detected using molecular biology techniques.

This anomaly is the most common among mutations affecting coagulation factors, with significant geographical variations: affecting around 5% of the population in northern Europe and the United States, its frequency decreases towards southern Europe, with frequencies of around 2% among Hispanics, 1% among African Americans and Africans, and 0.5% among Asians. The frequency of homozygotes has been estimated at 0.02%.[7]

Thrombosis occurs in deep veins in 60% of cases, in superficial veins in 27% of cases, and in arteries in 13% of cases.

Clinical symptoms vary depending on the location of the arterial thrombosis, its extent, and the effectiveness of collateral circulation.[8]

Doppler ultrasound is the most commonly used method for confirming the diagnosis of arterial thrombosis.

The treatment of these newborns involves two aspects: the first is medical treatment based on antibiotic therapy, with or without heparin therapy. The second is surgical, but given the major psychosocial impact on the patient and their family, as well as the tissue regeneration capacity of newborns.[7-8] We tend to delay treatment by not performing amputation immediately while waiting for a definitive demarcation of the gangrenous area, but in cases of established gangrene, amputation is the treatment of choice. There is no consensus on the indication for family screening. Nevertheless, screening asymptomatic carriers allows primary prevention measures for these individuals to be tailored to their family history of thrombosis in high-risk situations.[8]

However, there is no international consensus on the management of neonatal arterial thrombosis, and practices vary between centers.

Limitations

Because this study is based on a single case, generalizations cannot be made. Imaging and pathological evaluations were performed at a single center, which may introduce observer bias. Due to the limited number of cases in the literature, it is difficult to establish standardized diagnostic and therapeutic approaches.

Conclusion

The rarity of these neonatal arterial thromboses means that decisions are currently based on the practices of each individual team. In the absence of randomized studies, it is acceptable, subject to contraindications, to use thrombolytics whenever a thrombosis is extensive enough to threaten the patient's survival. The wide variety of clinical manifestations requires rigorous investigation by Doppler ultrasound to detect the presence of one or more thrombi and an assessment of the consequences in order to guide the associated therapies.

Ethics Declarations
The authors confirm that all procedures performed in this study were conducted in accordance with institutional ethical standards and the principles of the Declaration of Helsinki.

Animal and Human Rights Statement
All procedures performed in this study were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.

Informed Consent
Written informed consent for publication of the clinical details and images was obtained from the patient's parents/legal guardians.

Data Availability Statement
The datasets used and/or analyzed during the current study are not publicly available due to patient privacy reasons but are available from the corresponding author on reasonable request.

Conflict of interest
The authors declare that there is no conflict of interest.

Funding
None.

Author Contributions (CRediT Taxonomy)
Conceptualization: I.E., A.B.
Methodology: I.E., L.E.I.
Investigation: I.E., S.B.
Data curation: I.E., L.E.I.
Writing – original draft: I.E.
Writing – review & editing: A.B., S.B.
Supervision: A.B.

Scientific Responsibility Statement
The authors declare that they are responsible for the article's scientific content, including study design, data collection, analysis and interpretation, writing, and some of the main line, or all of the preparation and scientific review of the contents, and approval of the final version of the article.

Abbreviations
APC: Activated Protein C
CT: Computed Tomography
MRI: Magnetic Resonance Imaging
PT: Prothrombin Time

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How to cite this article:
Ilham Elouardighi, Lamya El Iaziji, Soukaina Batali, Amina Barkat. *Arterial thrombosis revealing a Factor V Leiden mutation: A Case Report. Ann Clin Anal Med 2026;17(Suppl 2):S196-199*